

A case series report of Chronic Recurrent Multifocal Osteomyelitis (CRMO) patients and it's etiologic relationship with MEFV Gene mutations

ABSTRACT

Background and Objective: CRMO is an inflammatory disease that occurs more often in children. The clinical manifestations are periodic fever, pain and bone lesions especially in long bones. Most Children show symptoms such as limping and tenderness in the affected areas. CRMO diagnosed after bone marrow biopsy and MRI rules out other diseases, such as bone tumors and bacterial osteomyelitis

Methods: Blood samples were taken from all patients diagnosed with CRMO from June 2011 until September 2018. The samples were screened for the 12 common pathogenic variants by an RDB assay (FMF StripAssay, Vienna lab, Vienna, Austria) according to manufacturer's instructions. Consequently, twelve common mutations consisting E148Q in exon 2, P369S in exon 3, F479L in exon 5 and M680I (G/C), M680I (G/A), I692del, M694V, M694I, K695R, V726A, A744S, and R761H in exon 10 were determined.

Result: 9 patients were enrolled in this study, eight were male. The most common involvement location was tibia and the least common location was zygoma, calcaneus and radius. Regarding the duration of the involvement, the mean duration of the involvement in male was 3.1 ± 1.3 years and in female was $1 \pm 0,0$ year and in total was 1.2 ± 1.2 years. 6 patients had only 1 involvement location, 2 patients had 2 involvements and 1 patient had three locations involved. Two patients had positive MEFV gene mutation, both of them were male. One patient had E148Q/Wt heterozygote mutation and the other had K695R/Wt heterozygote.

Conclusion: There was no meaningful relationship between the age onset of disease and sex. There was also no meaningful correlation between the MEFV gene mutations in patients and gender, age and location of involvement. There is a significant relationship between MEFV gene mutation and multiple locations of involvement and long duration of involvement

Keywords: MEFV Gene - FMF - CRMO